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Appendix 3

This appendix was part of the submitted manuscript and has been peer reviewed.
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Appendix to: Ito N, Peña AS, Perano S, et al. First Australian report of vitamin D-dependent rickets type I. *Med J Aust* 2014; 201: 420-421. doi: 10.5694/mja13.00220.

Appendix 3. Mutations of human CYP27B1

		gDNA	cDNA	Protein	Exon	Number of Family	Ethnicity of family
missense mutation	1	g.170G>T	c.170G>T	G57V	1	3	Chinese x3
	2	g.195G>T	c.195G>T	Q65H	1	1	Chinese
	3	g.866G>T	c.217G>T	G73W	2	1	Chinese
	4	g.954G>A	c.305G>A	G102E	2	1	Saudi Arabian
	5	g.969G>A	c.320G>A	R107H	2	3	Japanese x3
	6	g.984C>T	c.335C>T	P112L	2	1	Argentina
	7	g.1023G>A	c.374G>A	G125E	2	1	Japanese
	8	g.1584C>T	c.428C>T	P143L	3	1	Japanese
	9	g.1629T>C	c.473T>C	L158P	3	1	Chinese
	10	g.1646G>A	c.490G>A	D164N	3	1	Japanese
	11	g.1721G>A	c.565G>A	E189K	3	1	Polish
	12	g.1722A>G	c.566A>G	E189G	3	1	Swiss
	13	g.2287C>G	c.962C>G	T321R	5	1	Japanese
	14	g.2496C>A	c.968C>A	S323Y	6	1	British
	15	g.2525C>T	c.997C>T	L333F	6	1	Chinese
	16	g.2532G>C	c.1004G>C	R335P	6	1	Japanese
	17	g.2555C>T	c.1027C>T	L343F	6	1	Belgian
	18	g.2873C>T	c.1144C>T	P382S	7	1	Japanese
	19	g.2894C>T	c.1165C>T	R389C	7	1	Japanese
	20	g.2894C>G	c.1165C>G	R389G	7	1	Chilean
	21	g.2895G>A	c.1166G>A	R389H	7	8	White-USA, French Canadian, Black-USA, Belgian, Argentina, Danish, Norwegian, Turkish
	22	g.3244C>T	c.1226C>T	T409I	8	3	Philipino, Chilean-White, Chilean
	23	g.3304G>C	c.1286G>C	R429P	8	1	Black-USA
	24	g.3312C>T	c.1294C>T	R432C	8	1	Chinese
	25	g.3375C>T	c.1357C>T	R453C	8	1	Haitian
	26	g.3393C>T	c.1375C>T	R459C	8	1	Chinese
	27	g.3806T>G	c.1433T>G	V478G	9	1	British
	28	g.3848C>T	c.1475C>T	R492W	9	1	Chinese
	29	g.3863C>G	c.1490C>G	P497R	9	1	Polish
nonsense mutation	30	g.1963G>A	c.722G>A	W241X	4	1	Polish
	31	g.2511G>A	c.983G>A	W328X	6	1	Korean
	32	g.2607C>A	c.1079C>A	S360X	6	1	Turkish
	33	g.3265C>G	c.1247C>G	S416X	8	1	Norwegian
	34	g.3316G>A	c.1298G>A	W433X	8	1	Japanese
deletion mutation	35	g.48-60delCTGGGCGCCCGAG	c.48-60delCTGGGCGCCCGAG	E20fs	1	1	Chinese
	36	g.165delG	c.165delG	K55fs	1	1	Hispanic
	37	g.911delG	c.262delG	V88fs	2	11	French Canadian x7, Irish/French Canadian, White-USA x3,
	38	g.960-970delGGCCCGAGCGC	c.311-321delGGCCCGAGCGC	R104fs	2	1	Chinese
	39	g.1871delG	c.630delG	L210fs	4	1	White-USA
	40	g.1934delC	c.693delC	S231fs	4	1	White-USA
	41	g.3460delA	c.1442delA	E481fs	8	1	Chinese
	42	g.3328delG	c.1310delG	G437fs	8	2	Chinese x2
	43	g.3868delA	c.1495delA	R499fs	9	1	Moroccan
Insertion mutation	**44**	g.3341-3342insAC	c.1323-1324insAC	P442fs	8	1	White-USA
	45	**g.3343-3344insCCCACCC**	**c.1325-1326insCCCACCC**	**F443fs**	**8**	**20**	**Chinese x5, Korean x3, British X2, Turkish x2, French Canadian, Filipino, Polish, White-USA, Black-USA, Hispanic, Italian, White-Australian**
deletion-Insertion mutation	46	g.850-854delGGGCGinsCTTCGG	c.201-205delGGGCGinsCTTCGG	Q67fs	2	1	Black-USA
splice-site mutation	47	IVS1+2T>G (g.197T>G)	c.195+2T>G	V66fs	intron1	1	Turkish
	48	IVS2+1G>A (g.1036G>A)	c.386+1G>A	E130fs	intron2	1	Pakistani
	49	IVS3+1G>A (g.1746G>A)	c.589+1G>A	G197fs	intron3	6	Korean x4, Japanese x2
	50	IVS6+1G>T (g.2665G>T)	c.1136+1G>A	R379fs	intron6	1	Italian
	51	IVS7+1G>A (g.2945G>A)	c.1215+1G>A	T406fs	intron7	1	White-USA